

Polycystic Kidney Disease (PKD) Information



*For Research in Polycystic
Kidney Disease*

ENGLISH

PKD Types and Prevalence

Polycystic kidney disease has two hereditary forms: autosomal dominant (ADPKD), the most common of all life-threatening genetic diseases, and autosomal recessive (ARPKD), a relatively rare disease that often causes mortality in the first month of life.

With the presence of PKD, cysts develop in both kidneys. There may be just a few cysts or many, and the cysts may range in size from a pinhead to the size of a grapefruit. A normal kidney is the size of a human fist. When many cysts develop, the kidneys can grow to be the size of a soccer ball or larger and weigh as much as 38 pounds each (14 kilograms).

Cysts are sacs of fluid that cause the kidney to enlarge and can hinder its filtering ability. Cysts also squeeze on blood vessels, forcing the pressure to rise. Because of this, the first symptom of ADPKD is often high blood pressure. Other symptoms include fatigue, frequent urination, blood in urine, headaches, kidney stones, and urinary tract infections.

The National Institutes of Health (NIH) in the United States estimates that one in 10,000 to one in 40,000 babies have ARPKD. By contrast, the dominant form of PKD (ADPKD) affects one in 400 to one in 1,000 persons.

For example, in the United States, there are more persons with PKD than the combined number of those with cystic fibrosis, muscular dystrophy, hemophilia, Down's syndrome, sickle cell anemia, and Huntington's disease! In fact, 600,000 Americans and 12.5 million people worldwide are estimated to have PKD, making it the most common life-threatening genetic disease. It is two times more common than multiple sclerosis and 20 times more common than cystic fibrosis.

The Facts of Polycystic Kidney Disease

Because PKD is an inherited disorder, the dominant form of the disease (ADPKD) is passed from one generation to the next by only one affected parent. Each child of an ADPKD parent has a 50 percent chance of inheriting the disease.

ADPKD equally affects men and women, regardless of age, race or ethnic origin, geographic location or socio-economic status, and does not skip a generation.

Normally, each person is born with two kidneys, tucked under the rib cage in the back of the body on each side of the spine. Kidneys filter blood plasma and produce urine, whereby waste products are eliminated from the body. Without properly functioning kidneys, waste products build up in the blood, causing a toxic condition known as uremic poisoning.

Complications of ADPKD can be loss of kidney function, brain aneurysms, mitral valve prolapse, frequent kidney or urinary tract infections, chronic flank or back pain, pancreas or liver cysts, enlarged heart, kidney stones, groin or abdominal hernias, and diverticulitis of the colon.

More than 60 percent of individuals with PKD develop kidney failure, for which dialysis and kidney transplants are reasonable treatments. However, there is currently no known cure for PKD.

Three clinical tests can be used to diagnose a person with PKD: ultrasound, computed tomography (CT) and magnetic resonance imaging (MRI).

Fear, denial and ignorance of the disease are PKD's greatest obstacles. Current research demonstrates that a person with ADPKD may be able to play a major role in controlling the development of his or her disease with regular health-care maintenance, a good diet and regular exercise. The **PKD Foundation** works to advance all areas of PKD education and information.

Common PKD Symptoms

Information from the **PKD Foundation** should be reviewed with your physician if you have one or more of these common ADPKD symptoms:

High blood pressure

A family history of kidney disease

Heart problems or strokes

Kidney stones

Frequent urinary tract infections

Constant or intermittent pain in the back, side or stomach areas

Blood in the urine

ARPKD symptoms and onset vary considerably from infancy (often causing newborn death) to even, in some instances, early adulthood. Because the cystic kidneys have the inability to concentrate urine, ARPKD individuals produce large volumes of urine. Virtually all patients will develop high blood pressure, some will have a poor appetite and short stature, and approximately one-third will need dialysis or transplantation by 10 years of age. All ARPKD individuals also have a liver abnormality known as congenital hepatic fibrosis.

Current Research on PKD

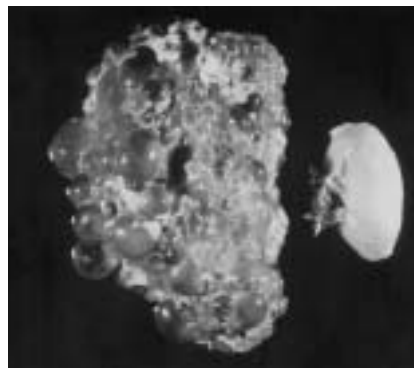
Genetic research is being done to determine the function of the PKD proteins—polycystin and polycystin-2—and to discover the gene for ARPKD.

Researchers are looking for new medications to help retard cyst growth and are devising diet strategies to control the development of PKD. Clinical research is being conducted to identify better methods of treating PKD and its complications.

A PKD registry of families is being gathered to analyze common family traits, which might isolate PKD causes and development. The International Gene Mutation Registry is cataloging mutations to see how they affect the progression of PKD.

PKD Foundation

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Normal kidney (right) compared to a polycystic kidney.

How to Handle PKD

The most important action to take is to learn as much as you can about PKD. Find a good kidney specialist (nephrologist). You can do this by getting a referral from your doctor or by contacting the department of nephrology at a major medical center nearest your home. Ask the head of the department to recommend a nephrologist or urologist familiar with PKD.

Keep your blood pressure under control. Check with your doctor for the proper medications and lose weight if necessary.

Watch your diet. Talk to your physician about counseling with a renal dietitian. Eating the right foods may help you feel better!

Avoid excessive amounts of caffeine—coffee, tea, colas, chocolate and some forms of aspirin (Anacin, Excedrin). Read labels!

Treat all urinary tract or bladder infections immediately. Contact your physician.

Do not use any over-the-counter medications without checking with your doctor. Avoid drugs with ibuprofen or naproxen (Advil, Medipren, Motrin, Nuprin, or Aleve).

Exercise your body. Staying fit by walking, swimming or light aerobics keeps your body healthy, lean and less prone to problems that can hasten the disease.

Our Mission

Once viewed as a hopelessly incurable disorder, polycystic kidney disease (PKD) has emerged as a prime target for study and treatment. The **PKD Foundation** is the catalyst for this growth.

We are the only organization, worldwide, devoted to programs of research to determine the cause, improve clinical treatment and discover a cure for PKD. We promote patient education, public awareness and advocacy for all PKD families.

We work to develop funding for peer-approved biomedical research projects. In addition, we engage committees and legislators within the United States Congress to promote the importance of PKD research conducted by the National Institutes of Health (NIH).

Our purpose is to bridge the gap between ignorance and knowledge about PKD by promoting programs of research and fostering awareness among medical professionals, corporations, foundations and the general public. We also provide the only regular source of PKD information, and we offer the only complete forum for patient education anywhere in the world through our annual conferences on PKD.